Chapter 6

The Evaluation of Anorectal Dysfunction in Paediatric Faecal Incontinence

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1. Introduction

Voluntary control of continence and defaecation is achieved during the first few years of development in children. Its acquisition can be delayed or sometimes secondarily lost through variations in the progression of developmental maturation, but also as a consequence of organic disease and dysfunction. The psychological impact of faecal incontinence can be devastating and yet it can be extremely difficult to treat effectively.

Although many factors are critical in the maintenance of continence, the anorectum is a remarkably complex organ which is the ultimate gate-keeper, mediating sensation of rectal content and allowing selective passage or retention of gas, liquid and solids. Functional anorectal disturbances which may lead to incontinence are mediated through sensory or motor nerves, smooth or striated muscle, the physical characteristics of the anorectum, or their conscious or unconscious coordination. The evaluation of anorectal function is an important component in the assessment of faecal incontinence at any age. There are a number of modalities available for the assessment of anorectal structure and function.[1] This review will review the role of methodologies for the evaluation of anorectal function with an emphasis on paediatric faecal incontinence.

2. Methodologies for the evaluation of anorectal function

The ideal functional assessment of continence would "provide dynamic information about the integrated function of each component, mimic situations where continence is threatened, discriminate between different treatment options and not interfere unduly with the normal physiological function of the organ."[2] With these aims in view the most commonly used techniques are described.

3. Anal tone and the pull-through technique

Anal sphincter tone can be measured by the pull-through of a sphere [3] or a balloon [4] by measuring the force required to pull it rapidly past the resting or consciously squeezing anal sphincter. Wald et al found that the force required to withdraw a solid sphere was similar in controls and children with encopresis.[3]

Shandling described using a balloon inflated with saline after insertion into the rectal ampulla. The maximum force resisting the rapid pulling out of the balloon is measured by a transducer. The proposed advantage of using a balloon in preference to a solid sphere was that there less likely to be an artefactual response by the sphincters after being stretched in order to insert the device. Meunier et al found that constipated children in general had significantly increased anal tone, but only one third (19/59) of the sub-group with encopresis showed elevation and this mean was not significantly different from that of control children.[5] In a later study of constipated children (44/63 with associated encopresis), the mean anal pressure was not significantly different from controls although 38% showed values greater than the normal range.[6] Loening-Baucke observed that rapid pull-through pressure was significantly lower in a group of 20 children with encopresis when compared with matched controls.[7] Moreover this failed to improve after successful laxative treatment. These pull-through techniques are now mainly of historical interest.

4. Saline continence

Threshold of incontinence to rectally infused saline can be used to measure the overall capacity of the anorectal unit in maintaining continence during conditions which partially simulate diarrhea.[8] The threshold volume at which leakage of at least 15 mL occurs is identified. Normally, in adult practice, the total volume retained is normally factored in as well. Most adults can retain 800 mL without leakage, but normal values for children have not been established. However, encopresis usually involves incontinence to solid stool and this method may be misleading.[4] Nevertheless, it continues to be used occasionally in adults where it can provide a useful measure.[9] It is of little value in paediatric practice.

5. Balloon expulsion

Expulsion of a rectal balloon, simulating defaecation, is used as an indication of the effectiveness of the defaecation mechanism and as a training device. A balloon attached to a narrow gauge probe terminating in a 3-way stopcock is inserted into the rectum and then inflated with

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a fluid such as water, either to a predefined volume (30-100 mL), or to sensory awareness.[8, 10, 11] The subject is asked to defaecate this within a prescribed time either seated on a toilet or in the left lateral position. Inability to evacuate the balloon within the specified time suggests the presence of anismus or dyssynergia.[8, 12] The technique may be modified by progressively reducing the distending volume in order to establish the volume at which evacuation does occur. This is generally a relatively well-accepted technique in children.

While saline continence testing and balloon expulsion provide simple screening tests for defaecatory disorders, they do not establish mechanisms.[13] They can however be used as repeated measures to define clinical progress.

6. Anorectal manometry

Manometric evaluation is the most established and widely available investigative tool for the evaluation of anorectal function.[1] It is widely used in paediatric practice, but generally requires a degree of cooperation and its utility may be limited, especially under the age of 5-6 years. Normal values obtained for children show a considerable variation depending on the technique (table 1).

Ref	N	% M	mean age (range)	ARM type	anal verge to balloon (cm)	rapid pull-through (mmHg)	resting anal tone (mmHg)	max. squeeze (mmHg)	RAIR (mL air)	transient sens'n (mL air)	prolonged sens'n (mL air)	total anal relax'n (mL air)	saline continence (mL)	balloon defaecation 100mL	anismus
[7]	20	75	8.6 (4-12y)	SS	11	133 (14)*	53 (12)	190 (49)	14 (5)						
[24]	15	66	8.5 (5- 12y)	SS	11				17 (7)	19 (7)	118 (29)	122 (46)+			
[43, 54]	16	69	10.3 (7-13y)	SS	11		67 (12)	140 (52)	11 (5)	14 (7)	101 (39)	104 (49)+	212 (47)	100%	0
[5]	32	41	8.2 (1-15y)	WP	7-15				17 (7.2)	17 (7.1)					
[6]	51	59	(4-14y)	WP	7-15	53 (9.7)**			median 15	median 20					
[55]	25	48	5.7 (2m-12y)	WP	7	median 56**	52 (7.1)		12 (4.0)						
[3]	21	52	9.6 (7-14y)	TB	5			81 (6.6)	24 (2.4)	14 (1.9)					2 (10%)
[22]	11	55	(7m-16y)	TB	7		29 (7.9)		22 (10.6)	32 (11.9)					

1 Figures: mean (SD). N=number in study, M=male. ARM=anorectal manometry. SS=solid state, WP=water perfused, TB=three balloon Pressure converted to mmHg where appropriate.

* anal resting tone=trough pressure at the maximum tone on rapid pull through-atmospheric pressure.

**anal resting tone=maximum rectal pressure-maximum anal pressure on rapid pull through.

RAIR=rectoanal inhibitory reflex

+If threshold not attained before prolonged sensation then it was deemed to have occurred at the next volume increment

Table 1. Anorectal manometric values in normal children.

Anorectal manometry can be used to define sphincter function, rectoanal reflexes, rectal sensation, rectal compliance and dynamic disorders of defaecation.

A manometric system includes four components – a pressure sensing device (probe), an amplifier or recorder for processing and storing signals, a display, and in more modern systems, software for data analysis. A variety of probes are available – these include solid state transducers, water perfused or sleeve catheters, and water or air filled balloon catheters.[8, 14] All catheters usually have a distensible end balloon. The three balloon system initially developed by Schuster [15] is not in common use in children.

The catheters are generally introduced with the patient in the left lateral position with hips and knees flexed without formal rectal preparation in the case of normal subjects, although they should be asked to empty their bowels prior to the test. No bowel preparation at all is required for very small children.[16] In the case of constipated or incontinent patients, it is advisable to wait at least 1 hour after disimpaction. The use of enemas for this procedure has been shown not to affect colonic motility or manometric parameters in healthy children[17] but it has been suggested that measurements recorded with an inadequately emptied rectum may be unreliable.[3] A digital rectal examination is always advisable to determine residual stool content and anatomy. Children who are unable to tolerate a digital rectal examination are most unlikely to accept catheter insertion.

In the case of either perfused catheters or those with transducers, pressure is measured at radially orientated directions around and/or along the catheter. Older methodologies involved distances of up to several centimeters between side-holes or transducers, but the recent move toward high definition manometry has resulted in the use of up to several pressure measuring points every centimeter. Typically, the measuring points at the extremes of the array are used to define "rectal" or abdominal pressure, and a pressure in air outside the patient. Within the array, closely spaced measuring points define the anal canal and sphincters, with overlap to allow for sphincteric movement.

There are advantages and disadvantages in each of the two systems – either water perfused or solid state transducer. Water perfused systems are cheaper, more robust, but suffer from the difficulties associated with water leakage. Solid state systems are much more accurate and convenient but also much more expensive and fragile. The physical presence of either catheter in the anal canal of this device must inevitably result in some degree of artefact, and this may be influenced by the diameter of the probe. Water leakage may also make simultaneous electromyographic (EMG) measurement using surface electrodes difficult. Both systems also suffer from the trade-off between obtaining radially-directed and longitudinal measurements at the same time. Fixed techniques without some sort of slow-station withdrawal may not accurately reflect asymmetric abnormalities in sphincteric pressure.

An estimation of the length of the anal canal and of anal resting and maximal squeeze pressure using the station or pull-through technique can be obtained by either manometric system.

Anal pressures obtained from station techniques are less than half those obtained by the rapid pull-through method, possibly due to sphincteric spasm.[7] The station method is generally preferred.[1] Anal pressures have been shown to vary significantly within individuals due to various cyclic activities.[18, 19] Age is also likely to have a substantial impact, especially in childhood. Cyclic variations in anal pressure may well be of importance in influencing propensity for soiling[20], but are practically difficult to measure in children and are not routinely measured in adults.

It has been suggested that anal hypertonia in constipated children may contribute to obstructed defaecation. Hypotonia was suggested as contributing to incontinence. Arhan et al compared resting anal and rectal pressure between constipated children (121/176 with encopresis) and a control group.[21] The mean internal anal sphincter and rectal tone pressures were significantly elevated in constipated children, but mean external anal sphincter tone was normal. Molnar subsequently similarly found that the anal canal tone was hypertonic in a group of constipated children (28/32=88% with associated soiling).[22] However, in contrast, using the station pull-through method, anal tone was found to be significantly reduced in chronically constipated children (96% with faecal incontinence) by Loening-Baucke et al.[23] This was observed to normalize in the children who recovered from constipation and soiling after treatment with laxatives as opposed to the rapid pull-through pressure which did not. [7]

Rectal filling during anorectal manometry is mimicked by the inflation of a balloon into the rectal medulla, generally with its distal point of attachment to the probe between 6 and 11cm from the anal verge. This can be used to determine the presence of the rectoanal inhibitory reflex (RAIR), define rectal sensation and as a measure of rectal compliance.[8]

Balloon distension is relatively simple to use in children, and allows an indication of internal anal sphincter, external anal sphincter (EAS), rectal and balloon pressures at rest, during voluntary squeeze and strain and rectoanal inhibitory reflex. Transient and stepwise inflation of the balloon in predetermined increments of between 5 and 30 ml allows the approximate measurement of threshold volumes of the reflex internal anal sphincter relaxation, compensatory EAS contraction, transient sensation, sensation of fullness and desire to defaecate. Normal values for these have been defined for children. Unfortunately, there are a number of differing protocols for rectal distension, each with implications for normal values which results in difficulties comparing the results from different investigators.(table 1)

The rectal sensory threshold can be difficult to define, as the duration of initial sensation on progressive stepped balloon filling tends to be very transient. As the balloon is progressively filled in a stepwise fashion, sensation becomes more and more prolonged and accompanied by a desire to defaecate. We have used sensation lasting longer than 30 seconds to define the rectal sensory threshold but find it difficult to help children distinguish between a sensory threshold and desire to defaecate. Loening-Baucke [24] suggested a minimum of 30 seconds for urge to defaecate and tried to distinguish between a sensation of fullness (or mild urgency) and also from transient sensation. Sun et al [25] distinguished between sensation of wind and a desire to defaecate. These sensations are obviously subjective and very hard to define for children.

The rectal sensory threshold appears to be elevated in children with constipation with or without associated soiling.[5, 6, 10, 22]. The threshold for sustained internal anal sphincter relaxation may occur before that of sensation.[22] This is clearly important in relation to the occurrence of faecal incontinence.

Using progressive stepwise balloon distension to identify the threshold for prolonged (30sec) urge to defaecate, Loening-Baucke found that children with faecal retention and soiling had a significant impairment in sensory thresholds.[10, 24, 26] This impairment persisted for up to

3 years in most (5 of 8) children despite apparent recovery from constipation and soiling after treatment with laxatives.[24]

The rectoanal inhibitory reflex (RAIR) is a relaxation of the internal anal sphincter occurring after distension of the rectum. Normally simultaneous contraction of the EAS and puborectalis muscle counters this and preserves continence. The reflex is absent in both Hirschsprung disease and anal achalasia.[14] There are some studies that would suggest that the distending threshold volume required to elicit this reflex is elevated in children with encopresis.

Meunier et al [5] found that the threshold for RAIR threshold was higher in 6% of constipated patients. Molnar et al [22] observed that 16% of constipated children (28/32=88% with associated soiling) had an elevated threshold. Neither value was significantly different from the control groups. Wald et al found that the threshold volume for eliciting the RAIR in encopretic children was not significantly different from that of controls.[3] This was confirmed by Loening-Baucke and Younoszai [23] in chronically constipated children (67/70 of whom soiled), although they also found that a given increase in rectal balloon volume produced a significantly smaller IAS relaxation in these children. This impaired relaxation persisted after treatment with laxatives regardless of outcome.[7] However, in a later study Loening-Baucke found a significantly elevated mean RAIR threshold in 97 constipated and encopretic children.[27] She expressed caution in interpretation of this result as the comparison control group had a lower threshold than that observed in a previous control group (see Table 1).

Over all it seems likely that at least some encopretic children do have an elevated RAIR threshold but this but this may relate to pre-existing rectal enlargement that increases the balloon distension required before the wall is stretched.

Squeeze pressures may be measured. It is useful to ask children to either "squeeze" or "strain" during manometric examination. This helps define anismus or dyssynergia. The usefulness of the actual resulting pressure value is less clear. Maximum squeeze pressure in children with encopresis was found by Wald [3] to be higher than in normal children, but not significantly so, and by Loening-Bauke [7] to be significantly lower.

Rectal compliance can be measured by intra-balloon pressures obtained during progressive, intermittent distension of a rectal balloon and comparing this with intra-balloon measured pressure during distension in air outside the patient. This provides a measure of the accommodation of the rectal wall which in turn depends on the visco-elastic properties of the wall and mobility of the adjacent viscera. Rectal compliance is decreased with age and inflammation of the rectum. A higher rectal compliance has been described in megarectum and faecal impaction. Urge incontinence may be related to reduced compliance.[28] Disturbed rectal compliance appears to be an important component of paediatric constipation, and does not seem to contribute to non-retentive faecal soiling.[29] Despite this, recovery from functional constipation in children and adolescents does not seem to be related to degree of abnormality in compliance.[30]

Balloon expulsion, though described previously, also forms part of the anorectal manometric study and can be combined with sphincter pressure measurements.

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Constipated children with encopresis are significantly more likely to have difficulty defaecating a water filled balloon and produce higher and more protracted elevations in intra-abdominal pressure than controls.[26, 27] There was no apparent distinction in these small studies between the patients able and unable to defaecate the balloons either clinically or in thresholds for RAIR, transient sensation and desire to defaecate, or with the accompanying increase in abdominal pressure. Children who are able to defaecate a balloon during the manometric study are significantly more likely to recover from constipation and soiling with laxative treatment than those who could not, and the difficulty evacuating a balloon does not appear to improve with treatment. Paradoxically, some children and adults have greater difficulty with smaller balloons than larger ones.[10, 31]

Robinson and Gibbons first identified paradoxical contraction of the anal sphincters (anismus) during attempted defaecation in children with encopresis.[32] This was present in 74% of children with constipation and in none in their control group. Subsequently, paradoxical contraction has been shown to coincide with an increase in EMG during straining in contrast with control children, who showed a reduction in EMG activity.[26] There is a strong association between anismus and the inability to defaecate balloons but none with the magnitude of the increase in abdominal pressure during straining.[26] Laxative therapy was not observed to improve anismus even when remission from constipation and soiling was achieved. Children with abnormal expulsion dynamics were more likely to have primary than secondary encopresis and more severe constipation (as evidenced by a palpable faecal mass in the abdomen). The three factors of severity of constipation, anismus and inability to defaecate a 100 ml balloon were found to be predictors of laxative treatment failure.[27] Recovery has been found to be more likely in children who were trained to relax the EAS during defaecation attempts than those who did not learn to do so. In those children who learnt to relax the EAS, 100ml balloon defaecation and transient sensation threshold improved in those who recovered from constipation and soiling but not in those who did not recover.[33, 34]

Megarectum is a term which has been used indiscriminately historically to describe significant rectal distension. For various reasons, the term “enlarged rectum” is better-used for those patients with a rectum which occupies greater than 0.61 of the recto-pelvic ratio. “Megarectum” should be limited to those in whom a significant abnormality is defined on anorectal manometry, pressure-volume curves, or rectal compliance investigations.[35]

Although anorectal manometry may be regarded as socially unpleasant and invasive, in general terms it is a low risk procedure. Colonic perforation has been described,[36] and clearly the procedure should ideally be carried out gently, with a cooperative patient, appropriately stopping if significant discomfort occurs.

Most children with encopresis have some form of manometric abnormality. Meunier[6] observed that 94% of their sample of constipated children, 70% of whom had associated soiling, had an enlarged rectum when compared with normal children and 97% of these children demonstrated some form of pathophysiology.

7. Ambulatory anal manometry

This provides a method for identifying spontaneous relaxations of the anal canal during prolonged more normal physiological activity.[37] A system of non-perfused microtransducers within a probe connected to a small portable recording box is inserted into the anal canal and rectum of the subject. A marker and diary enables the identification of manometric changes coinciding with events such as the feeling of urgency, sensation of or the passage of flatus, micturition, defaecation, sleep and postural changes. Parameters of motility can be compared directly but especially over time. Recording in the upright position is more natural than in the lateral position required by conventional manometry both from a physiological and a psychological viewpoint. This recording method is in its infancy and has yet to be reported with children. It does, however, potentially provide insights into periodic variations in rectal motor activity and variations in anal tone that might explain such conditions as nocturnal incontinence and proctalgia fugax.[38, 39]

It has been found that up to one third of children with retention and/or soiling are unable to perceive rectal dilation until IAS relaxation is well established.[22] This means that these children would have difficulty maintaining continence as the rectum fills. Knowing that multiple spontaneous IAS relaxations occur during the course of a day, each of these pose a threat to continence if, in the case of a child with chronic retention, the rectum is not empty, there is no compensatory contraction of the EAS and puborectalis. and the EAS response is inadequate.[40] Failure of compensatory contraction has been identified in adults with soiling.[40] This seems likely to be the basis of "overflow" soiling which is diagnosed in children.

8. Electromyography

Electromyographic (EMG) recording of electrical activity in the external anal sphincter can give a more accurate indication of tonic activity of this sphincter than interpretation of pressure differentials from anorectal manometry, particularly when the two sphincters are working in opposition. Two types of electrodes are in predominant use: 1) surface electrodes placed near the anal verge with the reference electrode on the thigh and 2) bipolar needle electrodes inserted into the striated muscle of the sphincter. Sponge electrodes, and a solid plug electrode (both placed within the canal) have also been employed. The needle electrode causes significant discomfort and only picks up signals from adjacent motor units. It is of limited clinical value in childhood encopresis. It is certainly not appropriate for use in the conscious child. Surface electrodes are much easier to use by comparison. One criticism of surface electrodes is that they not distinguish between external anal sphincter muscular activity and that of the abductor and glutei.[41] More recent modifications using multi-electrode arrays on a gloved index finger allow the extraction of information on innervation zones position and overlapping of motor unit branches of the puborectalis muscle and its electrophysiological properties.[42] This offers considerable advantage to a paediatric population.

Sphincteric EMG activity should reduce during straining, and increase during squeezing. When recording EMG activity during strain, the accompanying increase in intra-abdominal

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pressure should also be confirmed. A change of less than 25% in the baseline EMG is considered normal, while an increase in activity of greater than 25% is considered to indicate incorrect defaecation dynamics or dyssynergia [27, 43]. During voluntary squeeze the increase in electric impulse activity of the EMG should be not be accompanied by an increase in abdominal pressure.

9. Radiography

Plain abdominal radiography has been suggested as a tool to assess faecal retention in children and Barr et al developed a scale for this purpose.[44] Other scales have also been developed and although they tend to have good intra-observer correlation, they generally have poor inter-observer reproducibility.[45] Meunier et al using barium enema studies observed that almost all children complaining of constipation show radiological evidence of an enlarged rectum and bowel and so considered this procedure neither diagnostic nor contributory towards an understanding of its pathophysiology.[6]

10. Gastrointestinal transit time

Gastrointestinal transit time can be assessed by measuring the rate of movement of either a single group or sequentially ingested radiopaque markers through the intestine by either a single,[46] alternate daily or daily abdominal radiographs and/or their excretion by counting them in faeces.[47-49] Cruder measurements involve the mouth to evacuation time of solid recognizable foodstuffs or colored material.[50] One modification of this technique is the radionuclide transit study, which allows a better determination of sequential progress through the entire gastrointestinal tract,[51] and more recently the wireless motility capsule.[48] Importantly, radio-opaque markers, nuclear transit and wireless capsules all provide a measure of anorectal retention.

11. Anorectal biopsy

Although Hirschsprung's disease is normally diagnosed in infancy, the possibility that a child suffering from intractable constipation with or without soiling may have this disease should be considered.[52] If it is suspected, suction rectal biopsy should be undertaken to establish the absence of ganglion cells, the universally accepted standard to confirm the diagnosis of Hirschsprung disease.

Typically, the recto-anal inhibitory reflex is absent in patients with Hirschsprung disease. Failure to wait for resting conditions to become established, using inadequate balloon volumes and interference in the tracing of the IAS by contraction of the EAS can all lead to false positive diagnosis but it has been shown to have a sensitivity of 85% and a specificity of 100%.[53]

Anorectal manometry can play a role as a screening test for children especially over the age of a year, and guide assessment in patients with Hirschsprung disease who have had surgery. It can also assist management in those in whom sphincter myectomy or botulinum toxin are being considered.[14]

12. Conclusion

A number of methods exist for the evaluation of physiological function and dysfunction in the anorectum. Anorectal manometric testing is central but the others are also key. Despite the many tests available, there is still relatively poor understanding of normal function, and especially so in children.

Abbreviations

EAS	external anal sphincter
EMG	electromyography
IAS	internal anal sphincter
RAIR	rectoanal inhibitory reflex

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